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Day-by-Day: Averie's Story

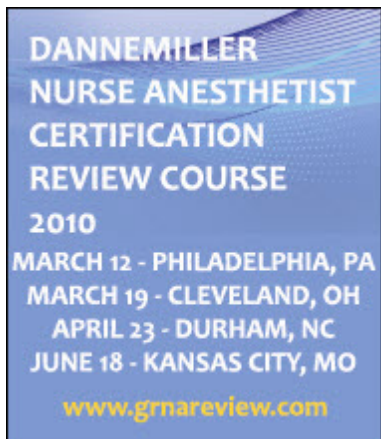
By Rebecca Rengo MA, MSW, LCSW, ACSW

In the middle of a busy classroom she sits alone. Kids are talking, laughing, and sharing food and notes. It's a party in kindergarten, then 6th grade, and now, high school. She remains silent. No one seems to notice or care, and she likes it that way.

Neurofibromatosis

Averie has Neurofibromatosis (NF) Type I. Although this condition is characterized by changes in skin coloring and the growth of tumors along nerves in the skin, brain and other body parts, Averie looks normal. NF occurs in one of every 4,000 births, affecting an estimated 100,000 people in the United States.

Between 30 and 50 percent of new cases arise spontaneously through mutation (change) in an individual's genes; many inherit the disorder as Averie did from



her father. If either parent has NF1, each child has a 50 percent chance of having the disease. Genetic counseling is recommended for anyone with a family history as the mutant gene can be passed on to future generations. Because of this condition, Averie feels different— like the ugly duckling trying to hide in a room full of people.

The First Signs

When she was two months old, Averie started having spots appear on her skin. Called café-au-lait spots, these are flat patches on the skin that are darker than the surrounding area. Her mother and family doctor noticed them, but didn't think they were significant. Many healthy people have one or two café-au-lait spots.

Doctors familiar with NF 1 look for six or more café-au-lait spots on the skin that are larger than five millimeters (mm) across in children, or 15 mm across in adolescents/ adults. In most people with NF 1, these spots may be the only symptom. Some of the other signs or symptoms may include:

- Blindness
- Convulsions
- Freckling under the arms or in the groin area
- Large, soft tumors (plexiform neurofibroma)
- Small, rubbery tumors of the skin (nodular neurofibroma)
- Pain from affected peripheral nerves
- Colored, raised spots called Lisch nodules on the colored part (iris) of the eye
- Mild cognitive impairment

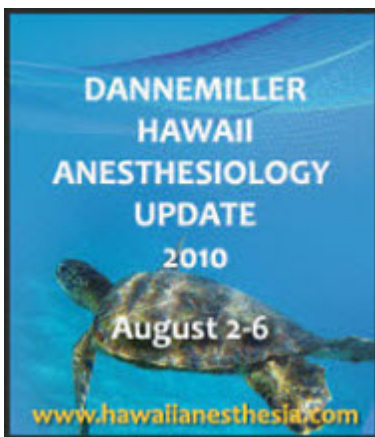
Children with NF 1 often have learning disorders. Diagnosis is usually based on the unique symptoms and signs of NF 1.

NF's Impact on Daily Life

At six, Averie fell off her bike and broke her arm. Her doctor did a work-up and referred her to a specialist. She had a tumor on the nerve of the bone in her arm. That's when her parents learned she had NF Type 1, also known as von Recklinghausen disease. Doctors told Averie she couldn't play sports and had to be very careful because she could easily break her arm again. She has worn a sling ever since.

Instead of childhood memories of sleepovers and sports, she remembers watching other girls giggle and play softball. Now 16, she overhears classmates at school discussing boys and getting their drivers' license. Meanwhile, Averie immerses herself in complementary and alternative (CAM) techniques for pain management. Aromatherapy, biofeedback, acupuncture... the list goes on and on. Her mother devotes her life to helping Averie manage her pain; she has taken training in Reiko, Emotional Freedom Technique and Cranio-Sacral therapies as most CAM techniques aren't covered under insurance. They all help. Using CAM as an adjunct to her pain medications keeps the pain at a "7 or 8;" she can function.

Associated Pain of NF



There are several different kinds of pain related to NF 1. According to Dr. Elizabeth Trinidad, pediatric neurosurgeon, "Some of them are structural, as with pain caused by scoliosis or mass lesions like neurofibromas that compress sensory neurons or other structures and can become malignant. NF1 patients should contact their physician any time there are symptoms such as:

- A rapidly growing tumor
- A previously painless tumor that becomes painful
- Weakness or numbness
- Changes in vision

When an NF1-associated problem is detected, there are often remedies that can improve the patient's quality of life or even be life-saving. Surgery can be performed to correct a curved spine, remove a tumor, or restore appearance. Sometimes chemotherapy and radiation can be helpful in association with surgery."

People with NF1 can experience more pain than normal from minor trauma, like Averie falling off her bike. Current research with mice is finding evidence that NF1 causes changes in the membrane structure that may make people with this disease more sensitive to painful stimuli. Research regarding this increased sensitivity is in very early stages. Treatment is targeted at managing symptoms.

Dorothy Stratman-Lucey, RN,MSN,BC,PNP, a Pain Management Coordinator & PeriAnesthesia PNP at Shriners Hospital for Children in St. Louis, MO, states, "We implement individual pain management plans for NF 1 patients. We educate parents and children; it's their right to expect quick response to pain. Our nurses are educated advocates. We ask all parents to attend an educational program to learn how to best care for their children's pain management needs. 96% of our patients and families attend these educational classes before surgery. Kids handle syringes, IVs, and other medical equipment to reduce their fears. Everyone is encouraged to ask questions. We tell them to expect to be listened to."

Stratman-Lucey goes on to say, "We use many non-pharmacological approaches as well. This helps to reduce opioid use. We use recreational therapy a lot, especially for younger children. For teenagers, we teach self-hypnosis. We use a variety of CAM approaches. I also stress the importance of addressing procedural pain (pain caused by shots and other procedures), including the technique used. Even if it takes extra time, it's well worth the effort."

Day-by-Day

Averie is stable; without any further complications, her life expectancy is almost normal. She doesn't think about her future. She tries not to think about her pain. Absorbed in crafts or cooking she has moments of pleasure. She's excited with a new camera and wants to study photography. "I try to do what I enjoy and just think about that. I don't worry about the future or dwell on the past."